

Aetiological investigation into severe to profound permanent hearing loss in children.

**Produced by the British Association of Audiovestibular Physicians and
British Association of Paediatricians in Audiology
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Aims

The aim of these guidelines is to update the evidence based approach to the investigation of the cause of deafness in children. Guidelines are 'systematically developed statements to assist decisions about appropriate care for specific clinical circumstances' based on systematic reviews of research literature (1, 2, 3).

Guidelines are not intended to restrict clinical freedom, but practitioners are expected to use the recommendations as a basis for their practice. Where possible recommendations are based on, and linked to the evidence that supports them. Areas lacking in evidence are highlighted and may form a basis for future research.

This is an update to the original BAAP/BACDA guidelines on aetiological investigation into severe to profound permanent hearing loss in children (2002)

Categories of evidence:

- Ia** Evidence from meta-analysis of randomised controlled trials
- Ib** Evidence from at least one randomised controlled trial
- IIa** Evidence from at least one controlled study without randomisation
- IIb** Evidence from at least one other type of quasi-experimental study
- III** Evidence from non-experimental descriptive studies, such as comparative studies, correlation studies, and case-control studies
- IV** Evidence from expert committee reports, or opinions or clinical experience of respected authorities, or both

Strength of recommendations is expressed thus:

- **A** directly based on category I evidence
- **B** directly based on category II evidence or extrapolated recommendation from category I evidence
- **C** directly based on category III evidence or extrapolated recommendation from category I or II evidence
- **D** directly based on category IV evidence or extrapolated recommendation from category I, II, or III evidence

Why investigate hearing loss?

There are several reasons why it is important to investigate deafness:

1. To try and answer parents who ask "Why is my child deaf? "
2. To identify and treat medical conditions e.g. 8th nerve aplasia congenital infection, Jervell and Lange-Nielsen syndrome, Alport's syndrome, Neurofibromatosis type 2, Ushers Syndrome, and vestibular hypofunction.
3. The results of investigations can assist the family in making decisions about the most appropriate communication mode, educational placement and counselling on cochlear implantation e.g. in 8th nerve aplasia, Ushers syndrome etc.
4. To inform genetic counselling
5. The information from investigation of childhood deafness informs epidemiological research

Subjects

All children with bilateral permanent hearing loss and thresholds over 70 dBHL in the better ear averaged across 500, 1000, 2000 and 4000Hz.

Guidelines for Good Practice

Level 1 investigations:

Level 1 investigations should be considered for every child. Timing will depend on several factors, including the family's agreement to proceed with tests, availability of local test facilities and how well the child can cooperate with tests.

In cases where aetiology has not been diagnosed then further aetiological investigations may need to be arranged and some repeated.

1) Paediatric history:

Detailed history of :

- onset of symptoms,
- pregnancy, delivery and postnatal period

- developmental milestones including speech, language, motor milestones as well as social development

History of exposure to risk factors e.g.

- noise
- ototoxic medications/ radiation
- head injury
- ear disease
- meningitis
- bacterial and viral illness
- immunisation status

Family history of deafness or risk factors associated with hearing loss in first and second degree relatives.

History of consanguinity and ethnic origin

2) Clinical Examination:

Should include height, weight and head circumference. Inspection of craniofacial region and physical measurements.

Examination of the ears, neck, skin and nails, limbs, chest, abdomen and gait.

Developmental assessment

3) Family audiograms: Parents and first degree relatives.(5)

4) Electrocardiography (ECG): for prolongation of the (corrected) QT interval, this is essential in children with evidence of vestibular hypofunction which may manifest as delayed motor milestones e.g. head lag, delayed sitting without support and walking. (6) (7). Please liaise with your local Cardiologist regarding the QTc interval norms. At Great Ormond Street Hospital is QTC of more than 460ms in girls and 450 ms in boys is considered as abnormal.

5) Ophthalmological assessment:

assessment of visual acuity and fundoscopy

discussion of electro-retinography with ophthalmologist if motor milestones are delayed (8, 9, 10, 11, 12) to detect Usher type 1

UNLESS child has adequate explanation for vestibular problems i.e.

Vestibular malformation

6) Urine examination (labstix) for microscopic haematuria (13, 14, 15)

7) *CMV screen (16, 17,18)

- < 1year of age

Urine CMV DNA PCR x 2 (separate occasions):

if positive, request Guthrie card for CMV DNA testing*

- >1 year of age

Urine CMV DNA PCR: and or IgG

if either positive, request Guthrie card for CMV DNA testing*

- Any age: consider testing mother's CMV IgG if not already known

If sending for Guthrie's consent from parents should be obtained

8) Blood test for Connexin 26 mutation (19, 20, 21, 22) with consent from parents, an explanation that DNA is stored afterwards in lab, that genetic testing can take a long time and permission to share results with other family members/professionals (see guidelines for consent for genetic testing (23)

9) MRI of Internal Auditory meati or CT Scan of Petrous Temporal bone (24, 25, 26, 27, 28, 29)

Level 2 investigations

Level 2 investigations will be indicated from history and clinical findings. As with level 1 investigations, timing will depend on the family's readiness to proceed with tests, availability of local test facilities and how well the child can cooperate with tests.

1) Serology:

to exclude congenital infection
to include maternal stored (booking) serum

2) Haematology and Biochemistry where clinically indicated

e.g. Thyroid Function tests indicated if:

- 1) Family history of thyroid disease
- 2) Goitre present
- 3) Widened vestibular aqueduct or Mondini deformity of Cochlea (30)

4) Investigation into autoimmune diseases where clinically indicated (31)

5) Metabolic Screen on blood and urine: where clinically indicated

6) Renal ultrasound:

- 1) If child has preauricular pits or sinuses, deformity of ear, branchial cleft or cysts
- 2) Mondini defect on imaging.
- 3) Permanent conductive or mixed hearing loss

7) Clinical photography**8) Chromosomal studies:**

History of developmental delay

Dysmorphic features

(Follow Child Development Team protocol)

9) Further genetic testing if indicated after discussion with the Geneticist

Consider referral to Clinical Geneticist especially

- if the parents are consanguineous,
- a syndrome is suspected,
- child has multiple problems,
- parental request
- opinion required on interpretation of genetic mutation testing
- After completion of investigations if a genetic disorder is diagnosed or no cause has been identified.

10) Vestibular investigations: Consider in all cases where motor milestones are delayed or where there is progressive deafness.

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